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1: Proc Natl Acad Sci U S A 2000 Jan 4;97(1):217-21

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Mutations in a NIMA-related kinase gene, Nek1, cause pleiotropic effects including a progressive polycystic kidney disease in mice.

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We previously have described a mouse model for polycystic kidney disease (PKD) caused by either of two mutations, kat or kat(2J), that map to the same locus on chromosome 8. The homozygous mutant animals have a latent onset slowly progressing form of PKD with renal pathology similar to the human autosomal-dominant PKD. In addition, the mutant animals show pleiotropic effects that include facial dysmorphism, dwarfing, male sterility, anemia, and cystic choroid plexus. We previously fine-mapped the kat(2J) mutation to a genetic distance of 0.28 +/- 0.12 centimorgan between D8Mit128 and D8Mit129. To identify the underlying molecular defect in this locus, we constructed an integrated genetic and physical map of the critical region surrounding the kat(2J) mutation. Cloning and expression analysis of the transcribed sequences from this region identified Nek1, a NIMA (never in mitosis A)-related kinase as a candidate gene. Further analysis of the Nek1 gene from both kat/kat and kat(2J)/kat(2J) mutant animals identified a partial internal deletion and a single-base insertion as the molecular basis for these mutations. The complex pleiotropic phenotypes seen in the homozygous mutant animals suggest that the NEK1 protein participates in different signaling pathways to regulate diverse cellular processes. Our findings identify a previously unsuspected role for Nek1 in the kidney and open a new avenue for studying cystogenesis and identifying possible modes of therapy.

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